

## Interesting case report ECARUCA (2015-02)

### Prenatal diagnosis of the duplication 17p11.2 associated with PotockieLupski syndrome in a foetus presenting with mildly dysmorphic features

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#### Abstract:

Duplication 17p11.2 (PotockieLupski syndrome (PTLS) MIM# 610883) is a genomic disorder with an estimated incidence of 1 in 25,000 births. As for other genomic disorders this duplication is typically de novo and is not associated with advanced maternal age or advanced paternal age. Herein we describe a prenatal diagnosis of duplication 17p11.2. This diagnosis was not suspected as the prenatal ultrasound findings were non-specific; however, BACs-on-Beads technology and array comparative genomic hybridization (aCGH) confirmed the common w3.7 Mb duplication. Evaluation of the foetus following termination of pregnancy revealed mildly dysmorphic features as well as congenital anomalies not previously reported in PTLs, specifically left pulmonary isomerism, an abnormally positioned left coronary orifice and nodular cerebellar heterotopia. This report exemplifies the utility of prenatal testing using new genomic technologies even when there are no multiple anomalies on foetal ultrasound. This report also exemplifies the utility of foetal autopsy in the identification of “occult” congenital anomalies.

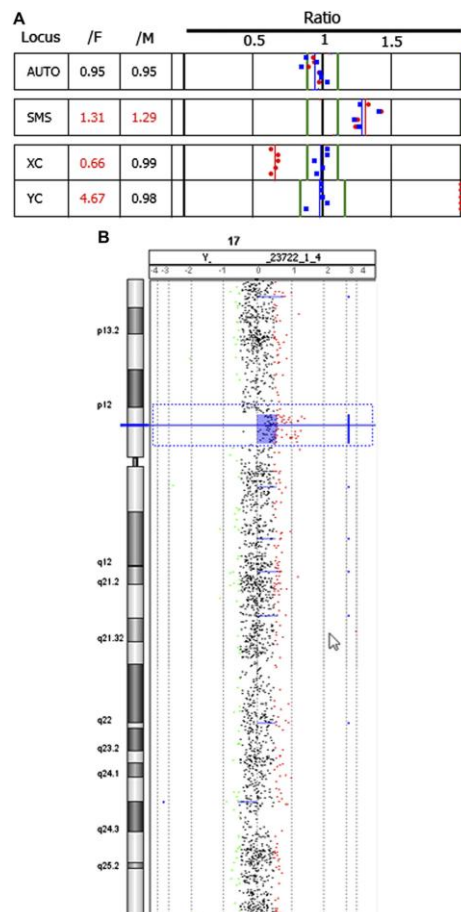


Fig. 1. Cytogenetic analysis. A: Prenatal BACs-on-Beads™ results for the male foetus with Smith-Magenis syndrome locus duplication. B: Confirmation of the 3.7 Mb 17p11 duplication by array comparative genomic hybridization.

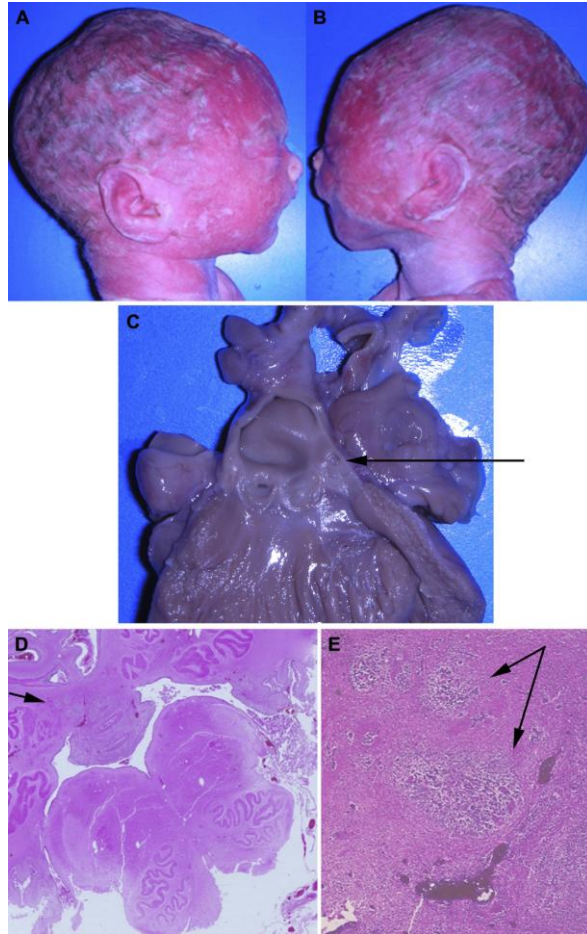


Fig. 2. Macroscopic and microscopic evaluations. A and B: Asymmetric ears. C: An abnormally high position of the (enlarged) left coronary orifice. D and E: small nodules of heterotopic neurons (D: x10, E: x100).